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Spondyloepimetaphyseal Dysplasia With Joint Laxity (SEMDJL): A Brazilian Case

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This is a report on a Brazilian patient with spondyloepimetaphyseal dysplasia with joint laxity (SEMDJL; MIM 271640), a rare autosomal recessive skeletal dysplasia characterized by dwarfism, articular hypermobility, progressive intractable spinal malalignment, a typical facies and a propensity to joint dislocation and subluxation. The condition has been described only in 20 children of Afrikaans-speaking parents in South Africa. This is the first report of a non-Afrikaans patient with this genetic entity.

KEY WORDS: bone dysplasia, dwarfism, joint laxity, autosomal recessive inheritance

INTRODUCTION

Beighton and Kozlowski [1980] described a unique form of spondyloepimetaphyseal dysplasia with joint laxity (SEMDJL) in seven South African children. Until now, only 20 patients were described with this new genetic entity, all in the Afrikaans-speaking population of South Africa [Beighton, 1994].

This is a report of a Brazilian patient with this rare condition and a discussion of the wider distribution of this rare gene.

CLINICAL REPORT

The patient is a 3-year-old Caucasian girl, who is the second child of a healthy 23-year-old mother and 39-year-old father. Ultrasonographic examination at 38 weeks of pregnancy showed shortened long bones and oligohydramnios. The baby was delivered by caesarian section with a birth weight of 3,100 g, length 45 cm, OFC 38.5 cm, chest circumference (CC) 33.5 cm; the Apgar scores were 8 and 9 at one and five minutes, respectively. Congenital dislocation of the hips and elbows and "hypotonia" were observed. Neuromotor development was normal, except that at age 3 the patient didn't have a normal gait because of articular laxity. The patient has normal mental development.

The parents are nonconsanguineous and clinically normal. The propositus is the second child of a sibship of two. The first child is a normal 7-year-old boy. Family history is unremarkable. The father has four healthy children of a first marriage. The mother has German ancestry. The family lives in Brazil.

At examination at 3 years and 3 months, the patient had short stature (79 cm, less than 3rd centile) weight 8,730 g (less than 3rd centile), OFC 50.5 cm (50–98th centile), chest circumference 51.5 cm (25–50th centile), relative macrocephaly, craniofacial disproportion, a typical facies of SEMDJL (oval face, blue sclerae, inner epicanthal folds, prominent eyes, low nasal bridge, and a long upper lip), high-arched palate, irregular teeth, short neck, thoracic asymmetry with pectus carinatum, severe kyphoscoliosis with lumbar kyphosis, severe articular hypermobility with elbow deformity (bilateral dislocation of the radial heads), unstable talipes equinovarus, terminal phalanges of the hands which are spatulate, and skin with a soft texture and some hyperextensibility (Fig. 1). The neurological examination was normal.

The radiological examination at 3 months (Figs. 2 and 3) showed gross spinal malalignment with dysplastic vertebral bodies (flattened and irregular); the radius was shortened and bowed with subluxation of the elbow joint, there were generalized metaphyseal widening and irregular epiphyses, large iliac wings with irregular acetabulae, short and broad femoral necks, and shortening of tubular bones of the hands. A cytogenetic examination was normal (46,XX).

DISCUSSION

The clinical and radiological picture of this Brazilian patient is very compatible with a diagnosis of SEMDJL (Table I). Torrington and Beighton [1991], in a genealogical study of the 13 nuclear families with 18 affected members with SEMDJL, showed the relationship of all the affected persons to two common ancestors (a founder effect in South Africa). Beighton [1994] said that "apart from the persons with SEMDJL in South Africa, the au-

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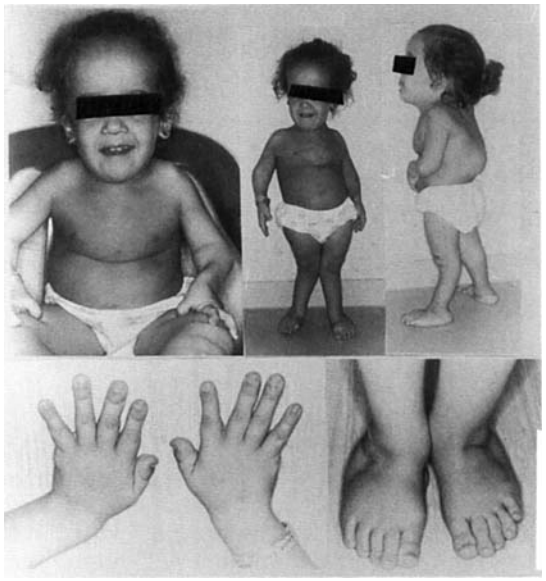

Fig. 1. The patient at 3 years and 3 months with relative macrocephaly, a typical facies of SEMDJL (oval face, prominent eyes, low nasal bridge, a long upper lip), short neck, pectus carinatum, severe kyphoscoliosis with lumbar kyphosis, elbow deformity, genu varum, some terminal phalanges which are spatulate, hyperextensibility of the joints, and unstable feet.

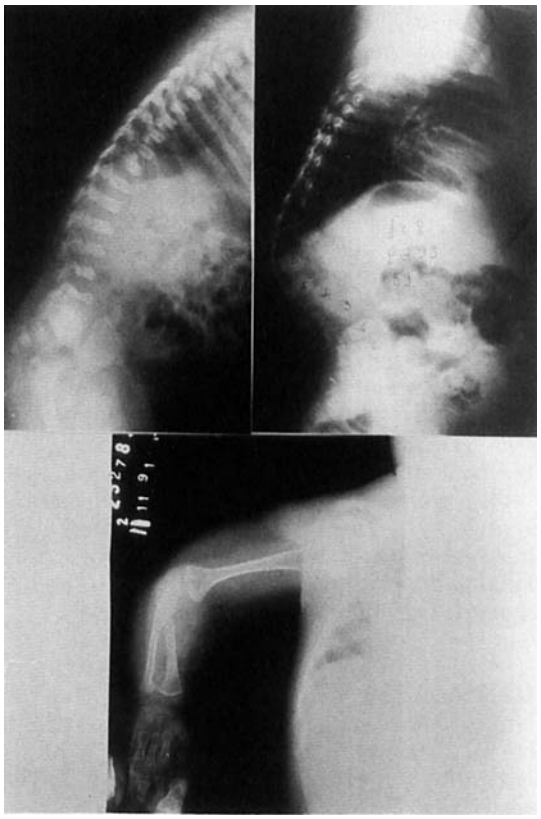

Fig. 2. Radiological manifestations at different ages: the vertebral bodies are flattened and irregular; the radius is shortened and bowed with subluxation of the elbow joint.

Fig. 3. Radiological manifestations at different ages: notable are gross spinal malalignment, large iliac wings with irregular acetabulae, short and broad femoral necks, shortening of tubular bones of the hands, and generalised metaphyseal widening and irregular epiphyses.

TABLE I. Clinical Manifestations of SEMDJL

	Present case
Consistent features present in all patients[a]	
Dwarfism	+
Articular hypermobility	+
Spinal malalignment	+
Thoracic asymmetry	+
Elbow deformity (bilateral dislocation of the radial heads)	+
Foot deformity (bilateral talipes equinovarus)	+
Characteristic but variable features	
Facies: Oval face	+
Long upper lip	+
Protuberant eyes	+
Scleral blueing	+
Hands: Spatulate terminal phalanges especially of the thumbs	+
Gross joint laxity permitting abnormal positioning	+
Skin: Soft, doughy texture with some hyperelasticity	+
Inconsistent features	
Cleft palate (31%)	−
High palate (12%)	+
Cardiac defect (28%)	−
Genu valgum (80%)	−
Congenital dislocation of the hips (27%)	−

[a]Beighton et al. [1984].

thor has been informed of affected persons in France, North America (Asiatic stock), and Germany (Turkish and German stock)." Herein we report a new case of this genetic entity not in Africa but born to Brazilian non-consanguineous parents. This reinforces Beighton's opinion that this gene is widespread in the world.

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